PHOTO QUIZ

An unusual cause of hyperammonaemia

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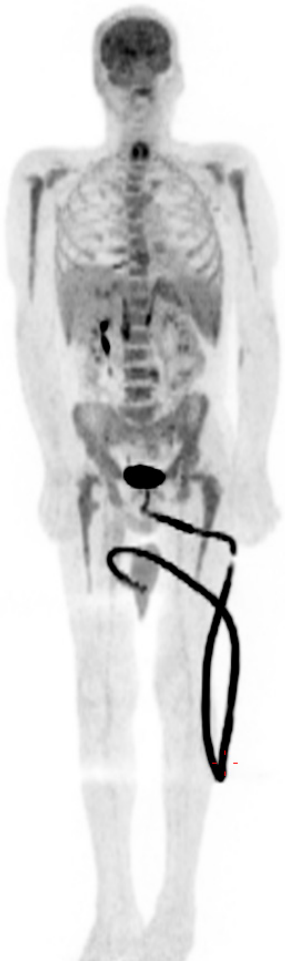
CASE REPORT

A 52-year-old man with a history of IL-12/23Rb1 deficiency presented to the emergency room with impaired consciousness. Laboratory investigations showed normal plasma glucose and electrolytes, but respiratory alkalosis, increased inflammatory parameters and pancytopenia. Lumbar puncture and head MRI did not show any abnormalities. An electroencephalogram revealed triphasic waves. Since these signs are often seen in metabolic encephalopathy, we checked plasma ammonia levels, which were elevated up to 216 µmol/l (reference 14-43 µmol/l). Liver function tests (i.e. clotting times and bilirubin levels) were normal. Moreover, abdominal ultrasound and CT scan excluded the presence of liver cirrhosis or flow anomalies. Blood, faeces, sputum and urine cultures and PCR all remained negative. Plasma amino acids (in particular glutamine), carnitine profiles and urinary orotic acid were all normal. A FDG PET-CT showed extensive FDG uptake in the bone marrow (*figure 1*). Bone marrow aspirate and biopsy were obtained (*figure 2*).

Figure 1. *Overview PET image shows physiological uptake in brain and bladder and diffuse bone marrow FDG uptake*



WHAT IS YOUR DIAGNOSIS?

See page 260 for the answer to this photo quiz

Figure 2. *Axial bone marrow biopsy stained by using the Ziehl-Neelsen method. Magnification x10*